



GLUTARIC ACIDEMIA, TYPE 1 (GA1)

FAMILY FACT SHEET

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 36 hours after birth. The blood is tested for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called GA1.

A positive newborn screen does not mean your baby has GA1, but it does mean your baby needs more testing to know for sure.

You will be notified by your primary care provider or the newborn screening program to arrange for additional testing.

What is GA1?

GA1 affects an enzyme needed to break down proteins from the food we eat, so they can be used for energy and growth. In GA1, the enzyme used to break down proteins is missing or not working properly.

A person with GA1 doesn't have enough enzymes to break down protein containing lysine and tryptophan, which causes a substance called glutaric acid to build up in the body and cause health problems.

GA1 is a genetic disorder that is passed on (inherited) from parents to a child. The mother and father of an affected child carry a gene change that can cause GA1. Parents usually do not have signs or symptoms, or even know they carry the gene change.

What problems can GA1 cause?

GA1 is different for each child. Some children with GA1 have few health problems, while other children may have very serious complications. Babies with GA1 are usually healthy at birth, but may be born with a large head.

If GA1 is not treated, a child might develop:

- Sleepiness
- Feeding problems
- Vomiting
- Muscle weakness
- Bleeding into the brain or eyes
- Cerebral palsy
- Seizures
- Coma

It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for GA1?

GA1 can be treated. Treatment is life-long and can include:

- Medications to help the body use protein and remove glutaric acid.
- Medications to help the body use energy.
- Diet low in lysine and tryptophan – a dietician will help you set up the best diet for your child.

Children with GA1 should see their regular doctor, a doctor who specializes in GA1, and a dietician.

Prompt and careful treatment helps children with GA1 live the healthiest lives possible.

Michigan Resources and Support

Michigan Newborn Screening

Nurse Consultant

Toll-free: 1-866-673-9939

newbornscreening@michigan.gov

Children's Hospital of Michigan Metabolic Clinic

Toll-free: 1-866-442-4662

Children's Special Health Care Services

Toll-free: 1-800-359-3722